



CARD14 gene

caspase recruitment domain family member 14

Normal Function

The *CARD14* gene provides instructions for making a protein that turns on (activates) a group of interacting proteins known as nuclear factor-kappa-B (NF- κ B). The NF- κ B protein complex regulates the activity of multiple genes, including genes that control the body's immune responses and inflammatory reactions. It also protects cells from certain signals that would otherwise cause them to self-destruct (undergo apoptosis).

The CARD14 protein is found in many of the body's tissues, but it is particularly abundant in the skin. NF- κ B signaling appears to play important roles in regulating inflammatory reactions in the skin and in promoting the survival of skin cells.

Health Conditions Related to Genetic Changes

familial pityriasis rubra pilaris

At least three mutations in the *CARD14* gene have been identified in people with familial pityriasis rubra pilaris, a rare hereditary skin condition. These mutations lead to overactivation of NF- κ B signaling, which triggers an abnormal inflammatory response in the skin. Researchers are working to determine how these changes lead to the specific features of familial pityriasis rubra pilaris.

psoriatic arthritis

other disorders

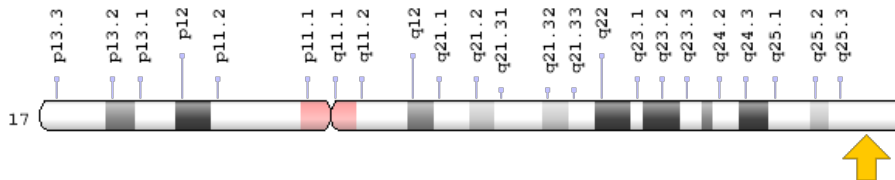
Changes in the *CARD14* gene may also contribute to a common inflammatory skin disorder called psoriasis. At least two mutations have been identified in families with psoriasis or psoriatic arthritis (a similar condition characterized by both skin and joint inflammation). Several other rare variants of the *CARD14* gene have been associated with an increased risk of psoriasis in people without a family history of the disorder.

Studies suggest that these genetic changes may enhance activation of NF- κ B, increasing the activity of genes involved in the body's inflammatory response. The resulting abnormal inflammation is a characteristic feature of psoriasis and psoriatic arthritis. However, *CARD14* gene mutations appear to be an uncommon risk factor for these complex conditions, which likely result from a combination of genetic and environmental factors.

Chromosomal Location

Cytogenetic Location: 17q25.3, which is the long (q) arm of chromosome 17 at position 25.3

Molecular Location: base pairs 80,169,992 to 80,209,331 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- bcl10-interacting maguk protein 2
- BIMP2
- CAR14_HUMAN
- CARD-containing MAGUK protein 2
- card-maguk protein 2
- carma 2
- CARMA2
- caspase recruitment domain-containing protein 14
- caspase recruitment domain family, member 14
- PRP
- PSORS2
- PSS1

Additional Information & Resources

Educational Resources

- Boston University: NF-kB Transcription Factors
<http://www.bu.edu/nf-kb/>
- National Institute of Arthritis and Musculoskeletal and Skin Diseases: Fast Facts About Psoriasis
https://www.niams.nih.gov/Health_Info/Psoriasis/psoriasis_ff.asp

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28CARD14%5BTIAB%5D%29+OR+%28%28BIMP2%5BTIAB%5D%29+OR+%28CARMA2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- CASPASE RECRUITMENT DOMAIN-CONTAINING PROTEIN 14
<http://omim.org/entry/607211>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_CARD14.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=CARD14%5Bgene%5D>
- HGNC Gene Family: Caspase recruitment domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/959>
- HGNC Gene Family: Membrane associated guanylate kinases
<http://www.genenames.org/cgi-bin/genefamilies/set/904>
- HGNC Gene Family: PDZ domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/1220>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=16446
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/79092>
- UniProt
<http://www.uniprot.org/uniprot/Q9BXL6>

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